



Scientific Updates on Organic Acidemias and Homocystinurias

Webinar | June 24-25, 2021

Thursday, June 24

Scientific session 1: Methylmalonic and Propionic Acidemias

10:00 - 10:05 a.m.	Introduction
10:05 - 10:30 a.m.	Genomic Therapies for Methylmalonic and Propionic Acidemia
10:30 - 11:00 a.m.	Liver Transplantation in Methylmalonic and Propionic Acidemia
11:00 - 11:30 a.m.	MMA and PA: First Guidelines Revision and Towards Novel Evidence
11:30 - 12:00 p.m.	Role of Mitophagy in Methylmalonic Acidemia
12:00 - 12:30 p.m.	Lunch Break
12:30 - 12:50 p.m.	Acyl Modifications in MMA
12:50 - 1:20 p.m.	 Mechanisms of Cardiomyopathy in Propionic Acidemia
1:20 - 1:40 p.m.	Liver Transplant and Cardiomyopathy in Propionic Acidemia
1:40 - 2:00 p.m.	Liver Neoplasms in Organic Acidemias: An Emerging Complication
2:00 - 2:30 p.m.	Break
2:30 - 2:50 p.m.	Machine Learning for Biomarker Data Mining in Organic Acidemias
2:50 - 3:10 p.m.	Hepatocyte-Based Models and Small Molecule Therapies for MMA/PA
3:10 - 3:30 p.m.	Neonatal Gene Therapy in Animal Models of Organic Acidemias
3:30 - 4:00 p.m.	Pave-GT Program for Organic Acidemias
4:00 - 4:30 p.m.	Panel Discussion

Charles P. Venditti, M.D., Ph.D.

Jerry Vockley, M.D., Ph.D.

Patrick Forny, M.D., Ph.D.

Olivier Devuyst, M.D., Ph.D.

Pamelasara Head, Ph.D.

Maria Fernandez-Velasco, Ph.D.

Gerard Berry, M.D.

Stephanie Grunewald, Ph.D.

Oleg Shchelochkov, M.D.

Kimberly A. Chapman, M.D., Ph.D.

Randy Chandler, Ph.D.

PJ Brooks, Ph.D.

Scientific Updates on Organic Acidemias and Homocystinurias

Webinar



Friday, June 25

Scientific Session 2: Combined Methylmalonic Acidemia and Homocystinuria

- 10:00 - 10:05 a.m. **Introduction**
- 10:05 - 10:30 a.m. Remethylation Defects
- 10:30 - 10:45 a.m. High-Dose B12 Therapy for cblC Deficiency
- 10:45 - 11:10 a.m. Pathophysiology of Manifestations of Methionine Synthesis Defects
- 11:10 - 11:35 a.m. Mouse Models of cblC, cblX
- 11:35 - 12:00 p.m. The Transcobalamin Receptor: Insights from Patients, Populations
- 12:00 - 12:30 p.m. **Lunch Break**

David S. Rosenblatt, M.D.

Irini Manoli, M.D., Ph.D.

 Jean Louis Gueant, M.D., D.Sc.

Ross A. Poché, Ph.D.

Lawrence Brody, Ph.D.

Scientific Session 3: Homocystinuria

- 12:30 - 1:00 p.m. Updates on Classic HCU Diagnosis and Management
- 1:00 - 1:30 p.m. Update on the Activities of the EHOD Collaboration
- 1:30 - 2:00 p.m. Newborn Screening for HCU
- 2:00 - 2:30 p.m. Treatment of CBS Deficient Mice by AAV-Mediated Gene Therapy
- 2:30 - 3:00 p.m. **Break**
- 3:00 - 3:20 p.m. Enzyme Replacement for Homocystinuria
- 3:20 - 3:40 p.m. How Elucidating Regulatory Cross-Talk Between Folate and Thiol Metabolism Can Lead to Improved Treatment in Classical Homocystinuria: A Possible Route to the End of Methionine Restriction?
- 3:40 - 4:10 p.m. Testing All Possible Amino Acid Changes in the Homocystinuria Proteins CBS and MTHFR
- 4:10 - 4:30 p.m. **Panel Discussion**

Harvey Levy, M.D.

Martina Huemer, M.D.

Marzia Pasquali, Ph.D.

Warren Kruger, Ph.D.

Tomas Majtan, Pharm.D., Ph.D.

Ken Maclean, Ph.D.

Frederick (Fritz) Roth, Ph.D.